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LATE-BREAKING ABSTRACT
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VALIDATION OF NON-INVASIVE GENE EXPRESSION (PLA) AGAINST HIGH RISK DRIVER MUTATIONS (BRAF, NRAS AND TERT) IN CUTANEOUS MELANOMA

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BACKGROUND: The diagnosis of early stage melanoma can be challenging histopathologically and can have a discordance rate as high as 27%. Non-Invasive gene expression testing for LINC and PRAME via a non-invasive pigmented lesion assay (PLA, 91% sensitivity, 69% specificity) has been validated against histopathology. Mutations in BRAF, NRAS and TERT promoter are found to correlate with melanoma tumor progression and histopathologic criteria.

TYPE OF STUDY AND METHODS: We sought to validate the PLA against key driver mutations in melanoma. A prospective/retrospective analysis of 103 PLA adhesive patch samples, with consensus panel confirmed histopathologic diagnoses, were analyzed for hotspot mutations in BRAF (non-V600E), NRAS, and TERT. Furthermore, a prospective analysis of mutation frequency in 523 real-world PLA samples was performed.

RESULTS: 97% percent of histopathologically confirmed melanoma samples were either PLA positive or mutation positive. Statistically significant differences in mutation frequency were observed between mel(+)/PLA+ and mel(-)/PLA(-) samples for hotspot mutations (75% vs. 15%, $p<<0.0001$). Mutations in adhesive patch samples were concordant with mutations in FFPE tissue blocks. TERT promoter mutations were the most prevalent (79%). Real-world PLA results showed that 89% of PLA(-) results were mutation negative, while 60% of PLA(+) results were mutation positive. There was no statistical difference in mutation frequency between validation samples and real-world samples.

CONCLUSIONS: This study confirms the high performance of the PLA. PLA positive tests identify high-risk lesions with driver mutations, while PLA negative test do not harbor these mutations. Gene expression and mutation analyses may enhance pigmented lesion assessment.

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